



lamellar ichthyosis

Lamellar ichthyosis is a condition that mainly affects the skin. Infants with this condition are typically born with a tight, clear sheath covering their skin called a colloidion membrane. This membrane usually dries and peels off during the first few weeks of life, and then it becomes obvious that affected babies have scaly skin, and eyelids and lips that are turned outward. People with lamellar ichthyosis typically have large, dark, plate-like scales covering their skin on most of their body. Infants with lamellar ichthyosis may develop infections, an excessive loss of fluids (dehydration), and respiratory problems. Affected individuals may also have hair loss (alopecia), abnormally formed fingernails and toenails (nail dystrophy), a decreased ability to sweat (hypohidrosis), an increased sensitivity to heat, and a thickening of the skin on the palms of the hands and soles of the feet (keratoderma). Less frequently, affected individuals have reddened skin (erythema) and joint deformities (contractures).

Frequency

Lamellar ichthyosis is estimated to affect 1 in 100,000 individuals in the United States. This condition is more common in Norway, where an estimated 1 in 91,000 individuals are affected.

Genetic Changes

Mutations in one of many genes can cause lamellar ichthyosis. These genes provide instructions for making proteins that are found in the outermost layer of the skin (the epidermis). The skin abnormalities associated with lamellar ichthyosis disrupt the normal formation of the epidermis, resulting in impaired regulation of body temperature, water retention, and resistance to infections.

Mutations in the *TGM1* gene are responsible for approximately 90 percent of cases of lamellar ichthyosis. The *TGM1* gene provides instructions for making an enzyme called transglutaminase 1. This enzyme is involved in the formation of the cornified cell envelope, which is a structure that surrounds skin cells and helps form a protective barrier between the body and its environment. *TGM1* gene mutations lead to severely reduced or absent enzyme production, which prevents the formation of the cornified cell envelope.

Mutations in other genes associated with lamellar ichthyosis are each responsible for only a small percentage of cases. In some people with lamellar ichthyosis, the cause of the disorder is unknown. Researchers have identified multiple chromosome regions that contain genes that may be associated with lamellar ichthyosis, although the specific genes have not been identified.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- collodion baby
- collodion baby syndrome
- ichthyoses, lamellar
- ichthyosis, lamellar
- LI

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 3
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1847849/>
- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 4A
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1832550/>
- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 5
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1858142/>
- Genetic Testing Registry: Autosomal recessive congenital ichthyosis 8
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3151377/>
- Genetic Testing Registry: Congenital ichthyosis of skin
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0020758/>

Other Diagnosis and Management Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST): Skin Care Tips
<http://www.firstskinfoundation.org/skin-care-tips>
- GeneReview: Autosomal Recessive Congenital Ichthyosis
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

General Information from MedlinePlus

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>

- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Lamellar Ichthyosis
<https://medlineplus.gov/ency/article/000843.htm>
- Health Topic: Skin Conditions
<https://medlineplus.gov/skinconditions.html>

Genetic and Rare Diseases Information Center

- Lamellar ichthyosis
<https://rarediseases.info.nih.gov/diseases/10803/lamellar-ichthyosis>

Additional NIH Resources

- National Institute of Arthritis and Musculoskeletal and Skin Diseases: Ichthyosis
https://www.niams.nih.gov/Health_Info/Ichthyosis/

Educational Resources

- Disease InfoSearch: Lamellar ichthyosis
<http://www.diseaseinfosearch.org/Lamellar+ichthyosis/9598>
- Foundation for Ichthyosis and Related Skin Types (FIRST): Lamellar Ichthyosis Type Fact Sheet
<http://www.firstskinfoundation.org/types-of-ichthyosis/autosomal-recessive-congenital-ichthyosis-arci-lamellar-ichthyosis-type>
- MalaCards: ichthyosis, congenital, autosomal recessive 1
http://www.malacards.org/card/ichthyosis_congenital_autosomal_recessive_1
- Merck Manual Professional Version: Ichthyosis
<http://www.merckmanuals.com/professional/dermatologic-disorders/cornification-disorders/ichthyosis>
- Orphanet: Lamellar ichthyosis
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=313
- The Swedish Information Centre for Rare Diseases
<http://www.socialstyrelsen.se/rarediseases/ichthyosis>

Patient Support and Advocacy Resources

- Foundation for Ichthyosis and Related Skin Types (FIRST): Lamellar Ichthyosis
<http://www.firstskinfoundation.org/skin-care-tips>
- National Organization for Rare Disorders (NORD)
<https://rarediseases.org/rare-diseases/ichthyosis-lamellar/>
- University of Kansas Medical Center Resource List
<http://www.kumc.edu/gec/support/ichthyos.html>

GeneReviews

- Autosomal Recessive Congenital Ichthyosis
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22lamellar+ichthyosis%22+OR+%22Congenital+Ichthyosiform+Erythrodermas%22+OR+%22Congenital+Ichthyosiform+Erythroderma%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Ichthyosis,+Lamellar%29+AND+%28lamellar+ichthyosis%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 1
<http://omim.org/entry/242300>
- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 3
<http://omim.org/entry/606545>
- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 4A
<http://omim.org/entry/601277>
- ICHTHYOSIS, CONGENITAL, AUTOSOMAL RECESSIVE 5
<http://omim.org/entry/604777>

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